



**European  
Reference  
Network**

for rare or low prevalence  
complex diseases

 **Network**  
Hereditary Metabolic  
Disorders (MetabERN)

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## SCIENTIFIC REPORT

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**1st POs MetabERN Meeting Frankfurt**

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January 13 2018  
Lindner Congress Hotel  
Frankfurt - GERMANY



# AGENDA

## MetabERN 1st POs MetabERN Meeting Frankfurt

13 January 2018, Lindner Hotel  
Frankfurt, Germany

### Morning session

- 10.00h** Welcome and general update by Maurizio and presentation of objectives and agenda of the day
- 10.10-10.40** Introduction round and main expectations of each of the participants
- 10.40-11.00** General overview of MetabERN and its goals – Christina Lampe
- 11.00-11.20** Overview of the work done by the Patient Board – Lut de Baer
- 11.20-12.15** First round of discussion and questions
- 12.15 -13.00** **Lunch**

### Afternoon session

- 13.00** Two parallel workshop sessions on the following topics:
1. Communication and cooperation among the Patient Representatives (PR) (webinars etc)
  2. Communication and cooperation between Healthcare Professionals (HCP) and Patient Representatives
  3. Setting up priorities: how is the collaboration with the HCPs in each country, patient needs and legal issues in different countries, access to therapy/home treatment, material for patients.
- 14.45** Feedback and report from the different sessions and discussion about the outcomes of parallel sessions
- 14.45-15.00** **Coffee break**
- 15.00-16.00** Closing remarks

## Participants





**European Reference Network**

for rare or low prevalence complex diseases

⊗ Network Hereditary Metabolic Disorders (MetabERN)

**Meeting 2018, January 13<sup>th</sup> – Patient Representation within MetabERN**

Participants	Patient Organization	Country	signature
Barbon Renza	ALSMITE ITALY-ePAG	ITALY	<i>Renzo Barbon</i>
Bellettato Cinzia	METABERN	ITALY	<i>Cinzia Bellettato</i>
Bond Simon	MetabERN	Italy	<i>Simon Bond</i>
Boonnak Julia	CDC UK	UK	<i>J. Boonnak</i>
Brown Colin	KRABBE UK	UK	<i>Colin R Brown</i>
Davidonis Martynas	Assoc. for ch. edwards rna dis.	Lithuania	<i>Martynas Davidonis</i>
De Baere Lut	BOKS vzw	Belgium	<i>Lut De Baere</i>
Dekker Hanka			
Devaux Marie	FEUX FOLLETS	FRANCE	<i>Marie Devaux</i>
Francisco Rita	Portuguese Association for CR	Portugal	<i>Rita Francisco</i>
Giuliani Margaret	VML	FRANCE	<i>Margaret Giuliani</i>
Hugon Anne	GSA (AFG)	France	<i>Anne Hugon</i>
Kosenko Lex			
Kuznetsov Mikhail	the road to life	Russia	<i>Mikhail Kuznetsov</i>
Lampe Christina		Germany	<i>Christina Lampe</i>
Lauridsen Anne Grethe	European Gaucher Alliance	DK/UK	<i>Anne Grethe Lauridsen</i>
Lavery Christine			
Meriluoto Anna	FINNISH FABRY ASSOCIATION	FINLAND	<i>Anna Meriluoto</i>
Owen Bill	NIEMANN-PICK UK	UK	<i>Bill Owen</i>
Scarpa Maurizio			
Sestini Sylvia	aim AKU	ITALY	<i>Sylvia Sestini</i>
Tobaldini Stefania	BAIAF Onlus	ITALY	<i>Stefania Tobaldini</i>
Da Campo Giovanni e Sara			
Torquati Fernanda	AIG FATORQUATI	Italy	<i>F. Torquati</i>
Van de Mheen Erica	Fabry Support & Information Group	Netherlands	<i>Erica Van de Mheen</i>
Wagner Leona	DSAKU e.V.	Germany	<i>Leona Wagner</i>
West Andrea			
MARIJA JOLDIĆ	MPS EUROPE		<i>Marija Joldić</i>

## Executive Summary

<b>Background</b>	<p>The MetabERN represents the first most comprehensive, pan-metabolic, pan-European, patient-orientated non-profit network established by the EU to facilitate access to the best available care and address the needs across the border of all patients affected by any rare inherited metabolic disease (IMDs) and their families.</p> <p>In particular MetabERN aims to connect the most specialized centers in the area of rare IMDs to <b>promote prevention, accelerate diagnosis and improve standards</b> of care across Europe for patients living with IMDs.</p> <p>MetabERN is driven by the principle of patient-centred care for the provision of its services aiming at improving the quality of life of patients and families. As stated in its mission MetabERN aims in fact to interact closely with patients, to serve as a reliable source of information and involve them in the planning of their individual care pathways and decision-making. So far MetabERN members have developed close collaborations with 44 national and European patient organisations (POs) and will build on their strong relationships to extend the networks engagement with the wider IMD patient community and break the isolation that many single patients currently experience.</p>
<b>Aims</b>	<p>The meeting held in Frankfurt aimed to give an overview presentation of MetabERN's objectives, to plan and define activities and programmes centred around high-quality patient care and to create a good basis for working together throughout the project.</p> <p>It offered patient representative (PRs) the opportunity to:</p> <ul style="list-style-type: none"><li>• directly discuss and understand the impact of your participation in the MetabERN and how patients can benefit from participation in the network;</li><li>• actively contribute to the empowerment of families associations;</li><li>• create synergies with other patients association in addressing common needs in the field of IMDs;</li><li>• have chance to exchange information during either the formal activities or the informal meetings with colleagues from different backgrounds.</li></ul>

### Opening remarks: aim of the meeting and network vision (Maurizio Scarpa)



Prof Maurizio Scarpa, the MetabERN Coordinator, opened the meeting and welcomed the participants. He gave an overall presentation of the agenda of the meeting and highlighted the importance of creating a collaborative team: knowing each other and starting collaborating at medical and patients level. *"Aim of the meeting, is for you (PR's) to tell us what to do"* stressed Prof Scarpa.

Today MetabERN involves 69 certified Health Care Providers endorsed by the Ministries of Health from 18 different EU countries. MetabERN is endorsed by and partners with the Society for the Study of the Inborn Errors of Metabolism (SSIEM). All these members aim to work together with POs in

a coordinated way to improve quality of life for people who are affected by certain rare metabolic conditions and their families.

The major goal of this initiative is in fact to **ensure a coordinated action in creating the widest collaboration among paediatric and adult metabolic physicians and patient associations at EU level, facilitating patient access to specialists with expertise in the metabolic field and to foster research activity.** MetabERN serves as a referral hub to ensure optimal knowledge-sharing, to improve prevention, early diagnosis and treatment of inherited metabolic diseases (IMDs) at EU level, to bring expertise at patient's bed, to facilitate access to therapy and to coordinate clinical and research services to rationalize the existing resources at European level.

It was particularly emphasized that **the patients and patients' empowerment are at the center of the interests of the MetabERN** and for this reason PRs will be involved in the different activities of the MetabERN, including the governance, ethics, care, research and evaluation of the MetabERN.

POs will then play a specific crucial role in the raising public awareness, dissemination of information about the diseases and their related treatment, and promotion of scientific research. They will also serve as a mediator between the patient and the professional.

## Introduction round and main expectations of each of the participants

During this session each participant introduces himself or herself shortly announcing his/her expectations about the meeting and the MetabERN in general.

Participants	Signature
Barbon Renza	It is difficult to understand the different patients' rights in the different countries but I think all <b>together we can do something very important and achieve strong results.</b> I'm here to actively work sharing my experience.
Boonnak Julia	Since we all have other different regular jobs it is hard sometime to find the time to dedicate to the different POs activities but <b>doing things together we can make our voice stronger</b>
Brown Colin	To meet as many people as possible and to collaborate together. <b>Working together we can strengthen patient empowerment</b>
Davidonis Martynas	To be able to <b>provide access to therapy to every patients affected by Fabry, to better understand the MetabERN and to better understand my role in it</b>
Devaux Marie	My expectation for today is to <b>satisfy my curiosity:</b> it's all about getting the picture and understand what needs to be done
Francisco Rita	<b>To establish connection and to promote patient centered research.</b>
Giuliani Margaret	<b>Be able to make the voice of small rare diseases and organisations loud.</b>
Hugon Anne	To <b>work together in a common effort,</b> sharing our own experience to <b>harmonize the knowledge and the way to contribute to meeting patients' need</b> and to <b>keep the voice of patients as a strong voice</b>
Maria Joldic'	<b>To gather all European organisations together. To meet patients' needs</b>
Kuznetsov Mikhail	<b>To make new connections and increase our network of support</b>

Lauridsen Anne Grethe	To connect name with faces and <b>finding out the mutual problems and what we can learn from each other</b> . To understand where I fit in and how I can help.
Meriluoto Anna	<b>Connect names with faces, to share knowledge learning from each other</b> , especially learning from other countries.
Owen Bill	I would like to understand more about the MetabERN and I hope it will enable quick and accurate diagnosis of patients and rapid access to treatment.
Sestini Sylvia	Mine is a small association and my hope is <b>to be able to make the voice of very small and very rare disease be present</b> . Differences from region to region are evident and since <b>each individual suffering from a rare disease is supposed to have the same right to the necessary treatments</b> I hope MetabERN can make something in this regard, at national and international level, <b>assuring that all the Italian and EU patients have the same quality and access to treatment</b>
Tobaldini Stefania	To share knowledge and experiences for the benefits of patients
Torquati Fernanda	We have a lot of things to do in Italy since we have so many problems to fight but I'm confident that <b>working together through MetabERN our voice can be heard</b> . In Italy we have class A citizens that can have home therapy and class B citizens that cannot. We would like <b>to explore and compare our local situation with other Countries in Europe</b> and understand who gets and who doesn't get home therapy. The hope for today is to be able <b>to make the voice of small rare diseases, and organisations loud and to gain home treatment for all patients</b>
Van de Mheen Erica	<b>To make a difference and do it better</b>
Wagner Leona	To <b>fight against the isolation rare disease patients often face</b> . To get and share Information on evidence based medicine, guidelines on diagnosis, new diagnostics testing, etc.

## General overview of MetabERN and its goals

(Christina Lampe)



Christina illustrated the specific aims of the MetabERN and the advantages for the patients community in having Patient representatives actively involved in its specific activities

### Major Aims of the MetabERN

- To pool knowledge and improve information exchange between network partners;
- To improve prevention, diagnosis and care in disease areas where expertise is rare;

### Major Opportunities Offered by The MetabERN to Patients

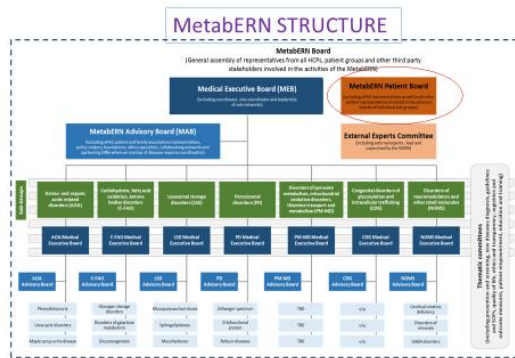
- To support Member States with a small number of patients to provide highly specialised care;
  - To advance innovation in medical science and health technologies;
  - To provide cross-border medical training and research
- 
- Optimization of care pathways for patients with rare disease: diagnosis, management access to therapy
  - More effective clinical trial designs that consider the very special and unique circumstances of rare disease research
  - Creation of databases/registries/natural histories of patients in a coordinated way
  - Close interaction with family associations which may help logistical challenges associated with conducting trials for small patient population
  - Close interaction among ERNs to integrate expertise needed for clinical trials
  - Possibility of helping Companies to generate new clinical trial designs based on the right choice of the primary and secondary endpoints based on the well-characterised patient population followed by the ERN.
  - Possibility of generating studies also for ultra rare patients due to the availability of numbers of patients in a coordinated way
  - Protection of patients from not well designed clinical trials
  - Counselling the patients in the understanding the value of the clinical trials Christina explained the structure and the activities of the MetabERN particularly highlighting the role and the objectives of the MetabERN patients Board.

### METABERN GOVERNANCE

The structure and the governance was shortly described, reminding to the MetabERN Website (<http://metab.ern-net.eu/>) for more details and information.

Prof Maurizio Scarpa, the Network Coordinator, is the legal representative of MetabERN. The Coordinator, assisted by the Medical Executive Board, supports and facilitates coordination within





and outside of the network. The Coordinator is assisted in the coordination activities by 3 nominated Vice-coordinators.

Stefan Koelker, Universitätsklinikum Heidelberg, DE

Eva M Morava-Kozicz, UZ Leuven, BE

Ans van der Ploeg, Erasmus MC, University Medical Center, Rotterdam, NL

The Chairperson of the SSIEM will also advise the coordinators.

**The MetabERN Patient Board (PB) and the MetabERN Advisory Board (MAB)**

support the MEB.

### The Medical Executive Board

The Medical Executive Board (MEB) acts as the implementation body of MetabERN. The MEB holds the strategic responsibility for activities of the network and will ensure its long-term sustainability. It consists of the **network coordinator, the 3 vice-network coordinators, the leaders of the work packages and the chairs of the horizontal work packages.**

MEB consults on a regular basis MetabERN's Patient Board and Advisory Board to provide input into the planning and implementation of activities.

### The MetabERN Patient Board

The MetabERN Patient Board is the **representation of all patient and family associations involved in MetabERN activities.** It is an independent body, which oversees and evaluates the work of MetabERN from the patients' perspective.

### The MetabERN Advisory Board

The MetabERN Advisory Board consists of representatives of the European Patient Advocacy Group, Patients and Family Associations, Foundations, Policy-makers, representatives from collaborating networks and other relevant stakeholders. Members serve for a 5-year renewable term.

An External Experts Committee (EEC), made up of HCPs not operating inside the MetabERN governance bodies, will also be set up to provide an external evaluation on the activities of the Network.

## MetabERN Structure

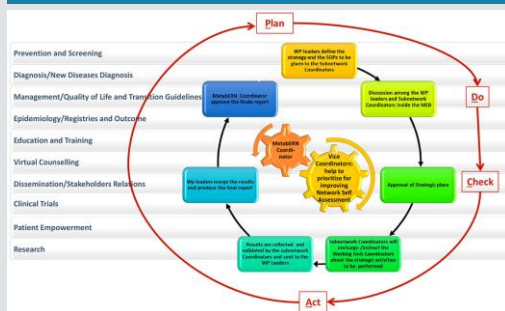
At its functional level, the MetabERN is composed of 7 Sub-Networks, each covering a different group of rare inherited metabolic diseases (see diagram above). A Subnetwork is led by 2 or 3 Coordinators who will elect their representative to sit on the MEB and MetabERN Board.

Each sub-group replicates at its level the structure of the overall MetabERN

See annex 1 for the list of the Subnetwork coordinators or consult MetabERN website (<http://metab.ern-net.eu>) for more details and information

The work programme of the MetabERN project is made up of 8 Work Packages (WPs). Each WP is led by one or two WP leaders See annex 2 for the list of the WPs leaders or consult MetabERN website (<http://metab.ern-net.eu>) for more details and information

## What are the roles of the Patient Associations inside the MetabERN?



Christina emphasised that the opinion of patient and family organisations will be taken into account for developing long-term activities and programmes centred on high-quality patient care.

Patients Association will be involved mainly in the:

### A: Governance of MetabERN:

by giving the opinion and views of patients and their families, advising on planning, assessing and evaluating the MetabERN, identifying and recommending expert centres and other HCPs that should join the MetabERN, establishing relations and mobilize relevant disease-specific POs when necessary.

### B: Overarching ethical principles:

to ensure that all ethical issues and concerns for patients are addressed, balancing patient and clinical needs appropriately, ensure transparency in quality and safety standards, clinical outcomes and treatment options; ensure compliance with the legal requirements on the protection of personal data, informed consent and sponsor-independency and transparency, making sure that patients receive relevant quality information, adapted to their health status, and that the principle of patients' ownership of personal data is respected, manage complaints for failure to comply with the above requirements

**C: Care:**

to ensure that care is patient-centred and respects patients' rights and choice, promote and encourage a patient-centric approach in the delivery of clinical care, service improvement, strategic development and decision making of the MetabERN, ensure the needs of all people with IMDs are considered and included in the MetabERN discussions and activities, specific to the scope of the MetabERN, contribute to the development and dissemination of information to patients, from policy to good practice, care pathways and guidelines, transparency and independency

**D: Research**

to contribute to the definition of research priority areas based on what is important to patients and their families, ensure that patients are embedded in the research activities performed within the MetabERN, including involvement in the assessment of clinical trials and ethics committees, contribute to the dissemination of research activities and outcomes, especially to patients that have been involved in research

**E: Evaluation:**

to ensure feedback and evaluation based on patient experience, reviewing the performance of the MetabERN by receiving and reviewing quality indicators, access times to diagnostic and treatment, clinical outcomes of diagnosis and treatment, evaluate of how the MetabERN acts based on feedback received from patients, though the ePAG, POs and patient experience surveys, in order to re-define and prioritize annually objectives, work plan and services

## How can the Patients Representatives actively contribute to the different MetabERN WPS?

To actively involve the PRs in the MetabERN Christina provided a clear overview of the different WPs explaining the specific tasks and the roles that PRs could play in each of them, underling that inputs and suggestions of task to be performed inside the WPs are really welcome.

**WP1\_Coordination**

This Work Package is developed for the good management of the project. The coordinator and the Coordination Team are the main responsible of this WPs

**Wp2\_Dissemination**

In parallel with WP1 this WP considers crucial the Interaction with EU institutions and the interaction with other ERNs. It was stressed that external dissemination is crucial to ensure wide dissemination of MetabERN's achievements. Dissemination must be specifically addressed two different target audience:

1. Dissemination of general information addressed to a lay audience and
2. ad hoc information for patients and their families as well as for healthcare professionals

**How can Patients Representatives actively contribute to this WP?**

- By directly participating in dissemination activities (giving presentations at conferences, publishing papers, networking and similar activities, with particular attention to acknowledge network affiliation)
- By exploiting opportunities to generate exposure of the project and the constant search for appropriate dissemination channels
- By helping the construction of a list of contacts which will be the recipients of the communication
- By periodically providing contents and news for the website, newsletter
- By contributing in the building, updating and maintaining of the website (explained step by step)
- By contributing in the Setting up and maintenance of the social media
- By developing general information addressed to a lay audience and ad hoc information for patients and their families as well as for HCP (Health Care Providers)

**WP3\_3 Evaluation**

This WP aims to make a robust internal evaluation of the MetabERN activities. Surveys will be instrumental for the main process.

**How can Patients Representatives actively contribute to this WP?**

- By directly assisting the Coordinator, Management Board and Advisory Board with monitoring progress
- By ensuring the interactions between stakeholders involved in the care for patients with IMDs are transparent and credible
- By evaluating the impact of MetabERN activities on patient care.

**Wp4\_ Guidelines, Care Pathways and standardization for medical care and for transition**

WP4 mainly concerns in the evaluation, into each subnetwork, of already existing GLs/CPs and in the provision of recommendations for the standardization for medical care and for transition

**How can Patients Representatives actively contribute to this WP?**

- By playing an active role in the process of standardization of recommendations, guidelines and care, assuring a patient friendly language.
- By playing an active role in the process of standardization of recommendations, guidelines and care, assuring a patient friendly language

### **WP5\_ Virtual Counselling Framework**

The Clinical Patients Management System (CPMS) is the main tool used by this WP aimed at advancing innovation in health technologies for IMDs. The CPMS is a secure web-based application provided by the EC to support European Reference Networks in the diagnosis and treatment of rare or low prevalence complex diseases or conditions across national borders. The system will allow for virtual consultation across national borders, ensuring that the needed expertise can travel to the patient, instead of the other way around.

#### **How can Patients Representatives actively contribute to this WP?**

- By using the Information Technology to inform patient community about the CPMS.
- By enabling patients to exert greater control over their own care.

### **Wp6\_ Research, Translational Activities & Clinical Trials**

Research is the main subject of this WP that aims at advancing innovation in medical science. Several issues in fact currently obstruct patient diagnosis and treatment including delayed diagnosis, limited availability of treatments and clinical trials for new treatments that are poorly designed and carried out. Identification of research needs and implementation of activities aimed at filling the gaps are the main focus together with the establishment of a grant office since lack of funding is another critical issue in research

#### **How can Patients Representatives actively contribute to this WP?**

- By taking part to group discussions with varying partners.
- Providing a coherent framework for further projects
- Facilitating the creation of cross-border multidisciplinary teams
- Facilitating the funding of research projects

### **WP7\_ Capacity-building & Training**

The main objective of this WP consists in increasing knowledge and skilling up competencies of target groups and supporting all Member States to provide highly specialised care to patients affected by IMDs

#### **How can Patients Representatives actively contribute to this WP?**

- Identification of training and teaching needs as well as already available capacities within the different Networks
- Develop a comprehensive program for the benefit of participating HCPs, young PhD students as well as patients and their families.

### **Wp8\_ Continuity of Care**

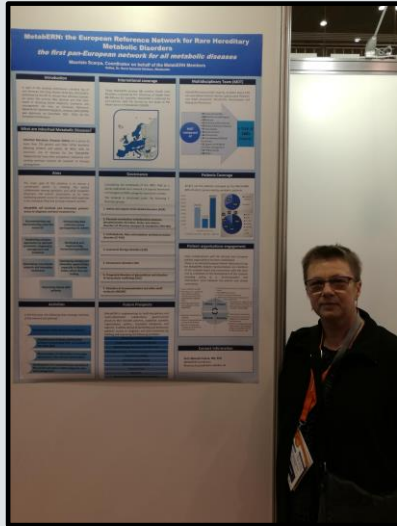
**Improving prevention, diagnosis and care in the disease areas covered by the Network is the main objective of this WP.**

#### **How can Patients Representatives actively contribute to this WP?**

- By taking part to group discussions with varying partners.

- By identifying via multiple channels those centers that could benefit from the MetabERN Network

## Overview of the work done by the Patient Board (Lut De Baere)



Lut De Baere, President of the Belgian Patient Organization for Rare Metabolic Diseases and responsible for the MetabERN Patients organizations (POs) management, stressed that the patients and patients' empowerment are at the center of the interests of MetabERN.

The main role of Patients representatives inside MetabERN consists in ensuring the widest patients involvement in the network. *"They must give a voice to every patient in Europe, breaking isolation that many single patients currently experience. Every patient in every European country, small or big must have access to equal care"* said Lut.

To achieve this goal it is crucial to identify and enrol POs representatives (PRs) that can act as "communication and information" actors, from professionals to patients, and vice versa. To this aim existing known POs at national, European and international level have been mapped and a formal proactive collaboration has been established with EURORDIS to facilitate the POs engagement and collaboration.

A preliminary list of some PRs to be involved in the different activities and taking part to the different working group inside each WP was shared. The file will be sent electronically to all the participants in order for them to be able to complete or amend it according to the specific competences.

Within the Patient Board an initial steering committee to start preliminary activities has been set up: Anne-Sophie Lapointe (Fr), Hanka Dekker (NI) & Lut De Baere (Be):

## First round of discussion and questions

### Letter of endorsement

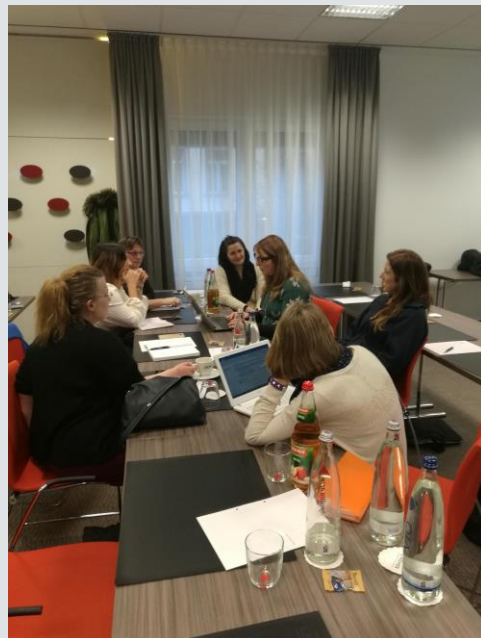
- PRs are requested to provide a letter of endorsement of their board. This is important because we must be sure that everybody is authorized by their board.

**Can it be translated?** Yes the letter can be translated in your own languages

## Two parallel workshop sessions



Due to the fact that some subnetworks were not greatly represented it was agreed to have two groups of discussion: one involving LSDs POs and the others



Participants were invited to participate in one of the 2 parallel breakout sessions according to their interest (LSDs or one of more diseases of the other 6 subnetworks)

Lut de Baere and Chirstina Lampe were

invited to guide discussion through the following major questions:

1. How is the general and daily care of metabolic patients organized in your country?

2. Do you have a Metabolic (Expertise) Center (1 or more; how many for how many inhabitants)?
3. Is the Metabolic Center (MC) for all metabolic diseases or do you more MCs for different diseases groups?
4. Is the care in that MC; multidisciplinary?
5. How is the access to diagnosis?
6. How is the access to care?
7. How is the access to expensive treatments (ODs, ..)?
8. How is the collaboration between PO and HCP?
9. Does the HCP inform the patient of the PO and his importance?
10. Do you have enough and updated material for patients (brochures)?



COUNTRY	LDS	OTHER 6 SUBNETWORKS
<b>1. <u>Do you have a Metabolic (Expertise) Center (1 or more; how many for how many inhabitants)</u></b>		
		•
BE		• 8 centers for 11 million people
DE		•
DK	• one National Center	• one for every province, 39 centers for 80 million people (most of them are for children)
FIN	• we have disease specific centers. We are treated, for example, as kidney patients, and the communication and attitude between hospitals and professionals hinders best possible care.	
FR	• Paris is the main center but we have 7 metabolic centers. Not sure if expertise is shared between local hospitals and them to help equal care for patients.	• 66 per 62 million people
IT	• we have a lot of centers but not all centers are expertise/reference centers, yet they say they are. Some reference centers only do blood tests. • Regarding Fabry (3 centers), there were two organisations (differentiated by 2 different treatments) they have now joined into one organisation.	• in theory we have 12-15 centers (one per each region) per 60 millions people
LT	• two centers but heart or kidney doctors treat patients, for example. Patients have to go from doctor to doctor.	
NL	• 3 specialized centers, that are well organized, but people don't always live close to the centers. We want local hospitals to be up to standard and be willing to search platforms in order to improve the care given to patients.	
PG	•	• 5 for 10 millions people
RU	• – a big center in Russia. Regarding care, when tests are run, the documents are sent to that center so care in the smaller towns is not adequate enough.	•
UK	• Scotland – two pediatric metabolic specialists, in Glasgow and Edinburgh.	• UK Yes, one in each major city, 14 centers for 65 million inhabitants

	<ul style="list-style-type: none"> <li>UK – 6 metabolic centers (pediatric and adult) Family goes to their local doctor and is then sent to a specialist.</li> </ul>	
<b>How is communication between Hospitals and PO's?</b>		
BE		the collaboration is very poor, because of competition among HCPs.
DE		<ul style="list-style-type: none"> <li>I guess that generally the collaboration is quite good and I found a lot of HCPs recommend the POs to pts. The collaboration has improved, the importance of POs has been recognized and the HCP inform the patient of the PO</li> </ul>
DK	<ul style="list-style-type: none"> <li><b>good contact</b>, the patient gets in contact with the PO or the nurse or physician ask the patient for permission to reach out to the PO's</li> </ul>	
FIN	<ul style="list-style-type: none"> <li><b>give information to the patient</b> and allow them to make the connection with the PO.</li> </ul>	
FR	<ul style="list-style-type: none"> <li>there is a <b>lack of communication</b> between hospitals and PO's</li> </ul>	<ul style="list-style-type: none"> <li>very good relation with HCPs, the HCP inform the patient of the PO</li> </ul>
IT	<ul style="list-style-type: none"> <li><b>they don't communicate with PO's</b>, and it is up to the patient to get in contact with the relevant Patient Organisations.</li> </ul>	<ul style="list-style-type: none"> <li>Communication is quite good, in Italy each HCPs have one POs.</li> <li>They are supposed to give information to the patient, but nobody value it</li> </ul>
NL	<ul style="list-style-type: none"> <li>Netherlands – <b>Quite good</b> have a folder/map with all information on disease, personal stories etc. in the hospital for when a person is diagnosed. POs are having meeting once or twice a year with physicians.</li> </ul>	
PG		<ul style="list-style-type: none"> <li>almost inexistent, doctors never inform, mainly because they lose time</li> </ul>
RU		<ul style="list-style-type: none"> <li></li> </ul>
UK	<ul style="list-style-type: none"> <li>UK and Scotland – they suggest a PO but the patient has to make the approach to the PO. Flyers and Info leaflets are left at info centers. Also not sure whether all patients get offered a PO by hospitals</li> </ul>	<ul style="list-style-type: none"> <li>poor, they do not really inform the POs</li> </ul>
<b>What is the patients' biggest need?</b>		
BE		<ul style="list-style-type: none"> <li>Improved access to therapy</li> </ul>

		<ul style="list-style-type: none"> <li>● Overcome the competition among different HCPs that has a negative impact on the Patients management</li> <li>● updated informative material for patients</li> <li>● Information from HCPs about POs</li> </ul>
DE		<ul style="list-style-type: none"> <li>● Overcome the lack of time of physicians</li> <li>● Overcome the bureaucracy</li> </ul>
DK	<ul style="list-style-type: none"> <li>● <b>communication</b> on all levels</li> <li>● <b>closer collaboration between PO's and Healthcare Professionals.</b></li> </ul>	
FIN	<ul style="list-style-type: none"> <li>● An equal procedure for all hospitals, like a <b>standard operations procedure.</b></li> </ul>	
FR		<ul style="list-style-type: none"> <li>● Awareness on IMDs especially using website</li> <li>● Training of general young practitioners</li> </ul>
IT	<ul style="list-style-type: none"> <li>● <b>clear protocol</b> for treating and following the patient and a reference centre has to be a centre with all the relevant information and capable of performing everything a patient might need to be done during a visit, for example, scans, blood tests, infusions, check ups etc.</li> </ul>	<ul style="list-style-type: none"> <li>● Overcome differences from region to regions different form region to region prenatal diagnosis</li> </ul>
LT	<ul style="list-style-type: none"> <li>● access to <b>home treatment</b></li> <li>● improve the awareness of rare diseases.</li> </ul>	
NL	<ul style="list-style-type: none"> <li>● make <b>medication available to everyone.</b></li> <li>● <b>Gender neutral.</b></li> <li>● <b>Early diagnosis.</b></li> </ul>	
PG		<ul style="list-style-type: none"> <li>● Information that is patients friendly, relevant and comprehensive</li> <li>● Improved access to therapy</li> <li>● Information form HCPs about POs</li> </ul>
RU	<ul style="list-style-type: none"> <li>● <b>improve the standards</b> around hospitals in Russia so that examinations can be done at your local hospital and don't have to be sent away, leading to long waiting periods for results.</li> </ul>	
RS	<ul style="list-style-type: none"> <li>● <b>information for patients</b> to find out more about their diagnosis etc. and <b>information for doctors</b> to improve their education on rare diseases.</li> </ul>	
UK	<p>UK and Scotland –</p> <ul style="list-style-type: none"> <li>● <b>improved newborn screening</b></li> </ul>	<ul style="list-style-type: none"> <li>● Improved access to therapy and newborn screening</li> <li>● informative material for patients and families</li> </ul>

	<ul style="list-style-type: none"> <li>● <b>better management plan</b>, for rare diseases, needs to be introduced (there is one but it's effect is yet to be seen).</li> <li>● <b>More information for Healthcare Professionals, regarding Rare Diseases</b>, to help them improve their knowledge.</li> </ul>	
<b><u>How is access to expensive treatment?</u></b>		
BE		it is difficult, complicated and nearly impossible
DK		
DE		<ul style="list-style-type: none"> <li>● depends form medical or public medical state. You have to know the health care system. Complicated</li> </ul>
FIN		
FR		<ul style="list-style-type: none"> <li>● for PKU the access is fine and don't expensive but I know it is not the case for all the other metabolic diseases. Diet and treatment for pku is free</li> </ul>
IT		<ul style="list-style-type: none"> <li>● depends form the governance of the regions, system is changing now, it is improving</li> </ul>
NL		<ul style="list-style-type: none"> <li>●</li> </ul>
PG		<ul style="list-style-type: none"> <li>● Not good bureaucratic and delayed</li> </ul>
RU		
UK		<ul style="list-style-type: none"> <li>● no access!!!</li> </ul>

What do we do next	
Things to be done	<ul style="list-style-type: none"> <li>● We need to identify the gaps,</li> <li>● We need to collaborate together to the organization of the meeting</li> <li>● We need to have POs involved in some voluntary work</li> <li>● We need to create the working groups and to this aim PRs needs to communicate us which WPs they are willing to contribute to</li> <li>● We need to define a steering committee of 4-5- PRs that are taking part in our main coordinating activities</li> </ul>

## MetabERN Board

(General assembly of representatives from all HCPs, patient groups and other third party stakeholders involved in the activities of the MetabERN)

### Medical Executive Board (MEB)

(including coordinator, vice-coordinator, leadership of sub-networks + leadership of WPs)

### MetabERN Advisory Board (MAB)

(including the SSIEM, ePAG patient and family associations representatives, relevant associations such as the European Metabolic Disorders Alliance, former and current policy-makers, foundations, ethics specialists, collaborating networks and partnering ERNs where an overlap of diseases requires coordination)

### MetabERN Patient Board

(including ePAG representatives as well as all other patient representatives involved in the advisory boards of individual sub-groups)

Sub-Groups

Amino- and organic acids related disorders (AOA)

Carbohydrate, fatty acid oxidation, ketone bodies disorders (C-FAO)

Lysosomal storage disorders (LSD)

Peroxisomal disorders (PD)

Disorders of pyruvate metabolism, mitochondrial oxidative disorders, thiamine transport and metabolism (PM-MD)

Congenital disorders of glycosylation and intracellular trafficking (CDG)

Disorders of neuromodulators and other small molecules (NOMS)

AOA Medical Executive Board

C-FAO Medical Executive Board

LSD Medical Executive Board

PD Medical Executive Board

PM-MD Medical Executive Board

CDG Medical Executive Board

NOMS Medical Executive Board

AOA Advisory Board

C-FAO Advisory Board

LSD Advisory Board

PD Advisory Board

PM-MD Advisory Board

CDG Advisory Board

NOMS Advisory Board

Phenylketonuria

N FRANK

Glycogen storage disorders

24

Mucopolysacchari-doses

Zellweger spectrum

TBD

n/a

Cerebral creatine deficiency

Urea cycle disorders

Disorders of galactose metabolism

Spingolipidoses

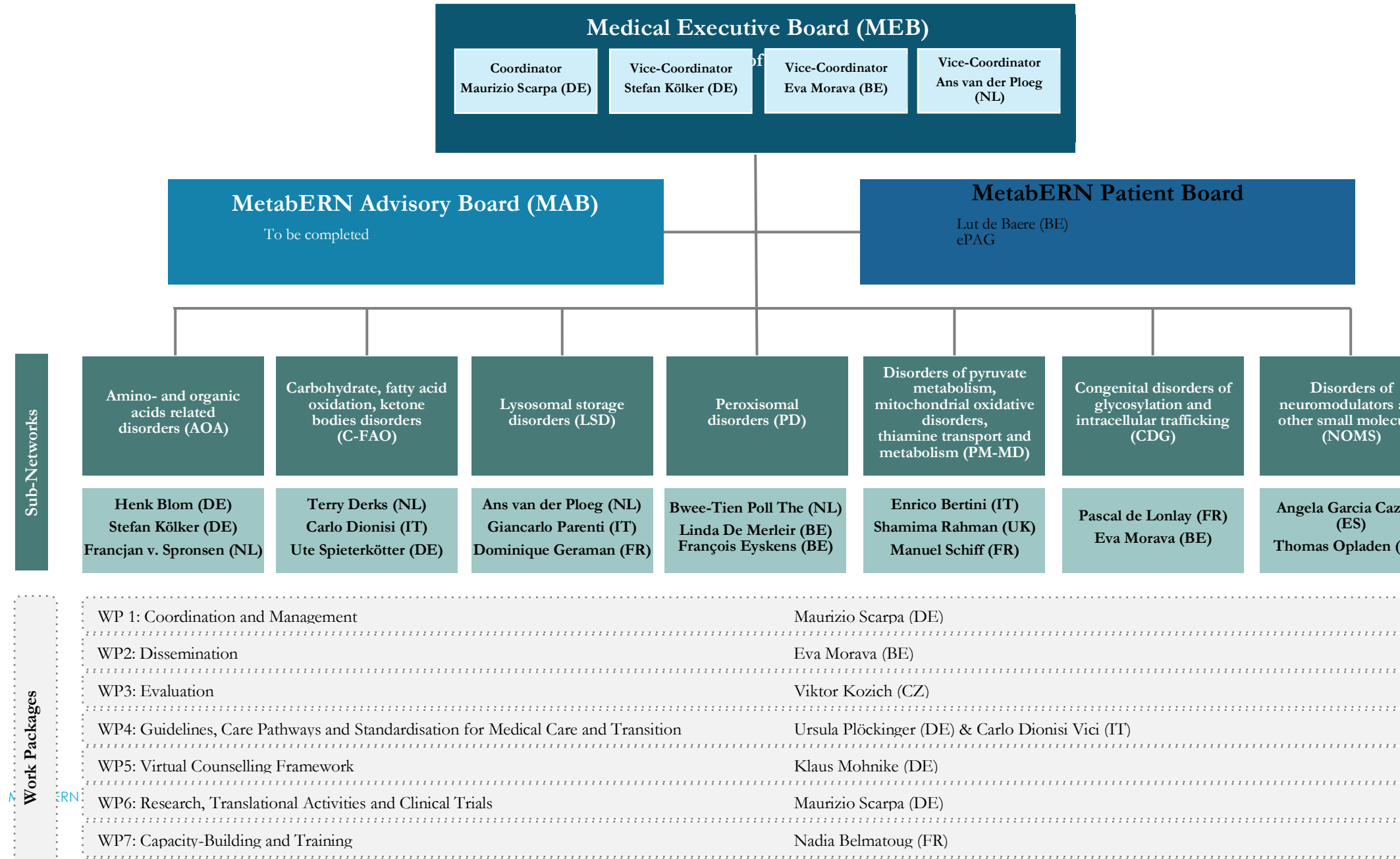
D-bifunctional protein

TBD

n/a

Neurotransmitter Disorders

ANNEX 2



## ANNEX 3

### VICE-COORDINATORS

<b>Vice-coordinators</b>	<b>Institution</b>
Stefan Koelker	Universitätsklinikum Heidelberg, DE
Eva M Morava-Kozicz	UZ Leuven, BE
Ans van der Ploeg	Erasmus MC, University Medical Center, Rotterdam, NL



## ANNEX 4

### SUB-GROUP COORDINATORS WITHIN THE METABERN

Sub-network	Coordinator
Amino and organic acids-related disorders (AOA)	<b>Henk Blom</b> <b>Stefan Kölker</b> <b>Francjan van Spronsen</b>
Disorder of pyruvate metabolism, Krebs cycle defects, mitochondrial oxidative phosphorylation disorders, disorders of thiamine transport and metabolism (PM-MD)	<b>Enrico Bertini</b> <b>Shamina Rahman</b> <b>Manuel Schiff</b>
Carbohydrate, fatty acid oxidation and ketone bodies disorders (C-FAO)	<b>Carlo Dionisi</b> <b>Terry Derks</b> <b>Ute Spieterkötter</b>
Lysosomal storage disorders (LSD)	<b>Ans van der Ploeg</b> <b>Giancarlo Parenti</b> <b>Dominique Germain</b>
Peroxisomal disorders (PD)	<b>Bwee-Tien Poll</b> <b>The Linda De Meirleir</b> <b>François Eyskens</b>
Congenital disorders of glycosylation and disorders of intracellular trafficking (CDG)	<b>Eva Morava</b> <b>Pascale de Lonlay</b>
Disorders of Neuromodulators and Other Small Molecules (NOMS)	<b>Angela Garcia</b> <b>Cazorla</b> <b>Thomas Opladen</b> (Porfira representative)

## ANNEX 5

### WP LEADERS WITHIN THE METABERN

<b>Work Packages</b>	<b>Leader</b>
WP 1: Coordination and Management	<b>Maurizio Scarpa (DE)</b>
WP2: Dissemination	<b>Eva Morava (BE)</b>
WP3: Evaluation	<b>Viktor Kozich (CZ)</b>
WP4: Guidelines, Care Pathways and Standardisation for Medical Care and Transition	<b>Ursula Plöckinger (DE)</b> <b>Carlo Dionisi-Vici (IT)</b>
WP5: Virtual Counselling Framework	<b>Klaus Mohnike (DE)</b>
WP6: Research, Translational Activities and Clinical Trials	<b>Maurizio Scarpa (DE)</b>
WP7: Capacity-Building and Training	<b>Nadia Belmatoug (FR)</b>
WP8: Continuity of Care	<b>Shamima Rahman (UK)</b>